

AperTO - Archivio Istituzionale Open Access dell'Università di Torino

Chiari and Syringomyelia Consortium: a model of multidisciplinary and sharing path for rare diseases.

This is the author's manuscript

Original Citation:

Availability:

This version is available <http://hdl.handle.net/2318/93547> since

Published version:

DOI:10.1007/s10072-011-0725-y

Terms of use:

Open Access

Anyone can freely access the full text of works made available as "Open Access". Works made available under a Creative Commons license can be used according to the terms and conditions of said license. Use of all other works requires consent of the right holder (author or publisher) if not exempted from copyright protection by the applicable law.

(Article begins on next page)



UNIVERSITÀ DEGLI STUDI DI TORINO

This is an author version of the contribution published on:
Neurological Sciences, Volume 32, Issue 3 Supplement, 2011,

doi: 10.1007/s10072-011-0725-y

Ciaramitaro P. et al., Volume 32, Springer, 2011, pp 271-273

The definitive version is available at:

<http://link.springer.com/article/10.1007%2Fs10072-011-0725-y>

Chiari and Syringomyelia Consortium: a model of multidisciplinary and sharing path for Rare Diseases

Palma Ciaramitaro , Simone Baldovino , Dario Roccatello , Edo Bottacchi •,Giuliano Faccani

Abstract Piemonte and Valle d'Aosta Interregional Network for Rare Diseases (RD) is a model of "diffuse" network; it involves all the health system specialists devoted to the diagnosis, the therapy and the follow-up of a RD. The Consortium is a multidisciplinary team operating throughout the Region composed of volunteer Physicians and Biologists that promotes periodical meetings to develop shared protocols. In 2008 the Specialist-Technical Committee for the Interregional RD Network approved the set up of the "Chiari and Syringomyelia Consortium" (CSC) with two specific objectives: to identify the shared diagnostic criteria and to develop the interdisciplinary diagnostic-therapeutic-assistance path (DTAP) to be used interregionally. Other objectives are: to define the prevalence of the pathologies (both symptomatic and asymptomatic, both in adults and children) and to improve collaboration between the patient Associations and the Physicians.

On behalf of the Interregional Piemonte and Valle d'Aosta "Chiari and Syringomyelia" Consortium.

P. Ciaramitaro , Clinical Neurophysiology Unit, AOU CTO, Turin, Italy

e-mail: p_ciaramitaro@libero.it

P. Ciaramitaro , G. Faccani

Neurosurgery Division, AOU CTO, Turin, Italy

S. Baldovino , D. Roccatello

Centro di Ricerche di Immunopatologia e Documentazione su Malattie Rare, Struttura Complessa a Direzione Universitaria di Immunologia Clinica Ospedale S.G. Bosco, Turin, Italy

E. Bottacchi

Department of Neurology, Ospedale Regionale, Aosta, Italy

Keywords Chiari malformation Á Syringomyelia Á Rare diseases Á Diagnostic-therapeutic-assistance path

Introduction

According to the European Union Actions Plans, the Rare Diseases (RD) are life-threatening, chronically disabling pathologies, with such a low prevalence requiring a specialists joint collaboration to prevent a significant premature mortality or morbidity, i.e. a relevant reduction in the quality of life or in the socio-economical potential of the affected persons [1]. In Europe the chosen threshold value was lower than one individual out of 2,000 to define a disease as Rare. With this criteria 5-8,000 different RD have been diagnosed out of 15 million of people in Europe alone. The diagnosis and care for RD has been indicated as priorities for Public Health by the Italian National Health Plan (1999); in 2001 a specific National Law (Ministerial Decree 279/2001) set up the Italian National Network for RD to deal with the prevention surveillance, the diagnosis and the treatment of the RD [2]. The same law activated the RD National Register located at the National Institute of Health (NIH), which is expected to receive epidemiological data from regional centres. The DM 279/2001 determines the RD that have right to ticket exemption, e.g. Arnold-Chiari Syndrome (code RN0010) for the symptomatic Chiari Malformation (CM).

Whilst the prevalence of Syringomyelia is known to be 8.4:100,000 [3], there are no available prevalence data for CM in Italy, because the case collection has started recently. A Regional Decree (2-03-2004, n. 22-11870) stated the Regional Network for the prevention, screening, diagnosis and therapy of Rare Diseases. Moreover it obliges the Regional Hospital Units to register RD in the Regional Network and sets up the Regional Centre for the Coordination of Rare Diseases [4]. A second decree (Regional Decree, 12-04-2005, n. 38-15326) sets up a Specialistic-Technical Committee to work in cooperation with the Regional Centre for the Coordination of RD and confers the benefits of the Ministerial Decree 279/2001 to about fifty RD [5], including Syringomyelia-Syringobulbia (exemption code RF0321). In fact, Syringomyelia is recognised as RD only in a few Italian regions, such as Piemonte, Valle d'Aosta, Toscana and Marche.

The Piemonte model of "diffuse" network is unique in Italy: it involves all the regional Health System specialists in the diagnosis, therapy and, above all, in the follow up and treatment. A crucial point of this model is carried out by the Consortium Activities, multidisciplinary groups, operating throughout the Region, composed by volunteer Physicians, Biologists and health care workers; they form working groups to enhance the research and the development of shared protocols for a standardized regional diagnostic-therapeutic approach.

In 2008, the Consortium Activities were approved with the aim of developing a dedicated, interdisciplinary and shared interregional diagnostic-therapeutic assistance path (DTAP); the next step will be to define the prevalence of Syringomyelia and Chiari in Piemonte and Valle d'Aosta, with special reference in the classification in symptomatic and asymptomatic classes, that may have a quite different outcome and consequent needs for health-care.

Materials and methods

The Interregional Piemonte and Valle d'Aosta CSC was promoted in Torino by Multidisciplinary Group for Chiari and Syringomyelia at CTO Hospital: this specialist team involves many branches of medicine and care (neurologists/neurophysiologists, neurosurgeons, neuroradiologists, physiatrists, spinal-surgeons, foniatrists, neuro-urologists, pain specialists, psychologists and social workers, dedicated nursing).

Its activities include the confirmation of the diagnosis and the patients selection for surgery, the planning of periodical Multidisciplinary Group evaluations both the surgical and non-surgical cases, the pre-surgical setup and post-surgical follow up and the identification of diagnostic-therapeutic protocols DTAP.

The DTAP begins with the neurologist in a dedicated neurological outpatients department and involves the other specialists in various extents, depending on the course of the disease. When a patient presents with suspected or defined Chiari Syndrome or Syringomyelia, then the diagnosis is confirmed by the neurological team that certifies it with an exemption request for RD. Instead, in the case of a preliminary diagnosis, a temporary exemption is supplied, to perform free diagnostic examinations further needed; after that, if the diagnosis is confirmed by the multidisciplinary team, the permanent exemption is given for free care and therapy. Whilst, if the disease is not confirmed, then the temporary exemption is not renewed.

The CSC organization has many further phases, which includes: preparing diagnostic-therapeutic criteria and differentiated protocols (symptomatic/asymptomatic; childhood/adulthood) as well as developing an interregional, shared DTAP.

Results

In a time span of a year, 102 patients were visited at the dedicated neurological outpatients department Chiari- Syringomyelia: 81 females, 21 males; 68 patients with radiological diagnosis of Chiari Malformation (65 CM-1 type and 3 CM-2 type; 37 isolated forms); 65 with radiological diagnosis of Syringomyelia (57 primary and 8 secondary forms; 26 isolated forms). Thirty-one patients were syringomyelia-Chiari I complex (30%). Forty-one patients (60%) with diagnosis of CM (Arnold-Chiari Syndrome, exemption code code RN0010) and 38 (58%) syringomyelic patients (syntomatic Syringomyelia, exemption code code RF0321) obtained the exemption, according to Ministerial Decree 279/2001 and Regional Decree, 12-04-2005. Meetings on Syringomyelia and Chiari were organized by CSC in Torino, Cuneo and Alessandria to share diagnostic-therapeutic criteria, to develop interregional protocols, to promote group research, to improve collaboration between patient Associations and physicians, to improve the relationships with the Italian Societies for Neurology, Neurosurgery and Rehabilitation.

Discussions and conclusion

Since 2005 in Piemonte and Valle d'Aosta there is a code for the patients affected by isolated Syringomyelia that avoids them from any expense for care or medication; whilst, the Chiari Syndrome patients have the complete exemption for RD in all the nation since 2001.

In Piemonte and Valle d'Aosta Interregional Network for Rare Diseases activated the CSC, "Chiari and Syringomyelia" Consortium, with the objective to obtain homogeneous diagnostic criteria, to develop a dedicated, interdisciplinary team and to share the same diagnostic-therapeutic paths in the two regions. Next step will be to define the prevalence of Syringomyelia and Chiari Malformation in Piemonte and Valle d'Aosta, with special reference to their clinical correlates (symptomatic and asymptomatic forms); moreover, it will organize health operators training programs, so enhancing the cooperation between the patients and the researchers.

The Interregional Piemonte and Valle d'Aosta "Chiari and Syringomyelia" Consortium:

Palma Ciaramitaro, Gianluca Isoardo, Paolo Costa, Clinical Neurophysiology, AOU CTO Torino, Italy; Giuliano Facchini, Michele Naddeo, Fulvio Massaro, Neurosurgery Division, AOU CTO Torino, Italy; Consuelo Valentini, Marilena Ferraris, Neuroradiology Division, AOU CTO Torino, Italy; Maria Vittoria Actis, Ilaria Rosso, Rehabilitation and Functional Recovery Division, AOU CTO Torino, Italy); Mauro Petrillo, Neuro-Urology Division, AOU CTO Torino, Italy; Stefano Aleotti, Antonio Bruno, Pasquale Cinnella, Spinal Surgery Division, AOU CTO Torino, Italy; Enrico Pira, General Medicine, AOU CTO Torino, Italy; Patrizia Consolino, Neurosurgery Division, CTO Hospital, Torino, Italy; Dario Roccatello, Simone Baldovino, Centro di Ricerche di Immunopatologia e Documentazione su Malattie Rare, Struttura Complessa a Direzione Universitaria di Immunologia Clinica, Ospedale S.G. Bosco, Torino, Italy; Paola Peretta, Pediatric Neurosurgery, Regina Margherita Children's Hospital, Torino, Italy; Alessandro Ducati, Marco Fontanella, Neurosurgery Division, Neuroscience Department, Torino, Italy; Enzo Luparello, Neurosurgery Division, Ospedale S.G. Bosco, Torino, Italy; Sergio Duca, Neuroradiology Division, Koelliker Hospital, Torino, Italy; Lorenzo Pinessi, Salvatore Gallone, Innocenzo Rainero, Neurosciences Department, AOU S. Giovanni Battista, Torino, Italy; Dario Giobbe, Neurology Division, AOU S. Giovanni Battista, Torino, Italy; Maria Pia Schieronì, Rehabilitation Division, S. Giovanni Battista, Torino, Italy; Maurizio Gionco, Headache Centre, Department of Neurology, Mauriziano Hospital, Torino; Federico Maria Cossa, Neuromotor Rehabilitation Unit, Casa di Cura Maior, Torino, Italy; Giovanni Asteggiano, Neurology Division, ASL CN2 Alba-Bra, Italy; Alessandro Mauro, Neurology Division, IRCCS Piacavallo-Neurosciences Department, Torino, Italy; Claudio Geda, Neurology Division, ASL Ivrea-Cirio-Chivasso, Italy; Luca Ambrogio, Neurology Division, Cuneo, Italy; Roberto Cantello, Clinical Neurophysiology, Neurology Division, Novara, Italy; Pietro Versari, Gianantonio Spena, Neurosurgery Division, Civil Hospital, Alessandria, Italy; Claudio Bernucci, Neurosurgery Division, Cuneo, Italy; Carlo Scamoni, Neurosurgery Division, Novara, Italy; Fabrizio Pisano, Maria Tommasi, Rehabilitative

Neurology Division, S. Maugeri Foundation, Veruno (NO), Italy; Edo Bottacchi, Department of Neurology, Ospedale Regionale, Aosta, Italy.

References

1. Comunità Europea. Decreto n. 1295/1999/CE, 29 aprile 1999. `Programma d'azione comunitaria sulle malattie rare nel quadro dell'azione nel settore della sanità pubblica (1999-2003). Gazzetta ` Ufficiale delle Comunità Europee 22 giugno 1999, n. 155
2. Italia. Decreto Ministeriale 18 maggio 2001, n. 279. Regolamento di istituzione della rete nazionale delle malattie rare e di esenzione dalla partecipazione al costo delle relative prestazioni sanitarie, ai sensi dell'articolo 5, comma 1, lettera b), del DLvo 29 aprile 1998, n. 124. GazzettaUfficiale - Supplemento Ordinario 12 luglio 2001, n. 160
3. Orphanet Report Series. Prevalence of Rare Diseases: bibliographic data. Novembre 2010, number 1, p. 27 http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf
4. Piemonte. Deliberazione della Giunta Regionale 2 marzo 2004, n. 22-11870. Bollettino Ufficiale della Regione Piemonte 1° aprile 2004, n. 13
5. Piemonte. Deliberazione della Giunta Regionale 12 aprile 2005, n. 38-15326. Bollettino Ufficiale della Regione Piemonte 19 maggio 2005, n. 20